



LUCID GENETICS

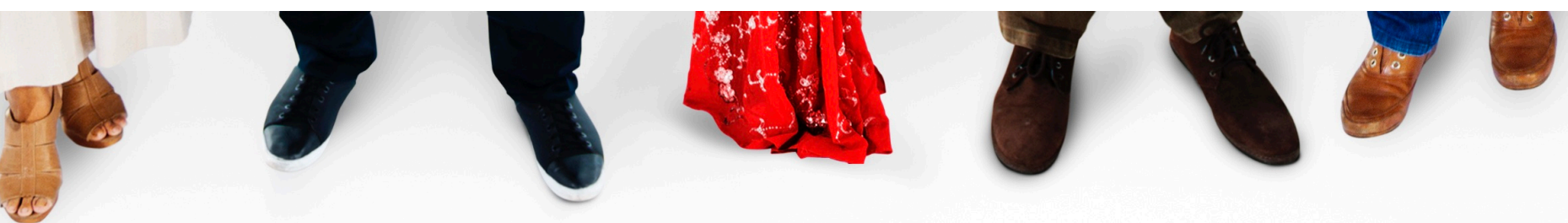
A TELEGENETICS MEDICAL PRACTICE



DOES IT RUN IN MY FAMILY?

A GUIDE TO FAMILY HEALTH HISTORY, GENETICS AND HEALTH

GENETIC ALLIANCE -- A FREE PATIENT EDUCATION BOOK



DEDICATION

This ebook is given at no charge.

This patient education book was created as an open source ebook by Genetic Alliance, Inc. (501c3).

Lucid Genetics, Corp. a telegenetics medical practice, updated the artwork and layout but the content has not been altered.

We encourage you to share this book with family and friends; the book is free and not copyrighted. We hope this benefits those who are looking for answers and offers an understanding to this new era of medicine -- genetics. This book is the beginning, please keep researching and learning about this field because it involves all of us.

Leigh J Mack, MD, PhD, FAPCR, CPI

President

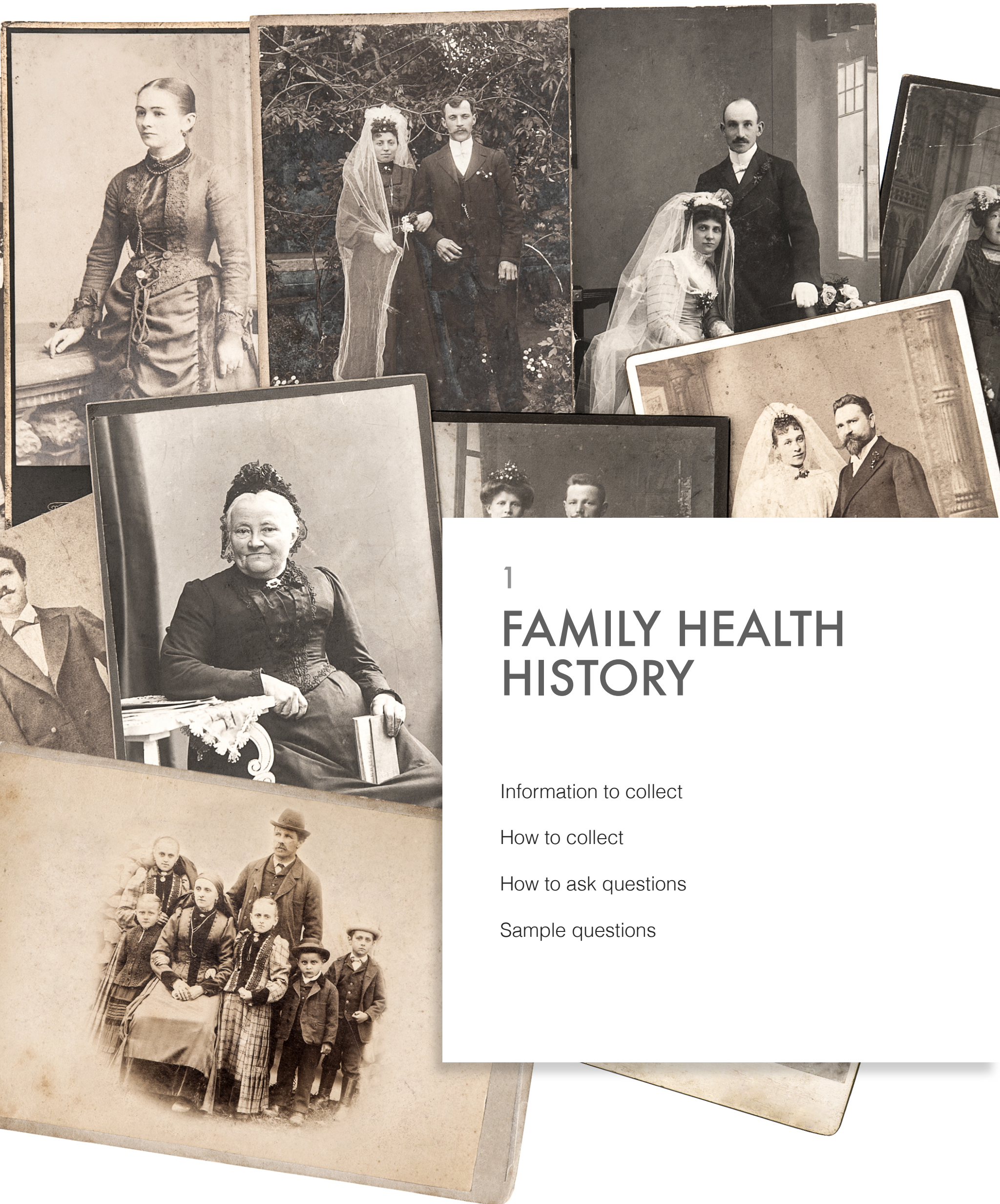
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1 FAMILY HEALTH HISTORY

Information to collect

How to collect

How to ask questions

Sample questions



COLLECT

What is family health history?

Family health history is information about diseases that run in your family, as well as the eating habits, activities, and environments that your family shares. Knowing about the diseases that run in your family can help you make healthy choices.

Your family's health is one part of the history of your family. While collecting your family health history, pay attention to events, stories, and experiences as well. Gathering your family history helps you share your family stories and health information with your family members and children.

“ Family health history can be found in the choices you make, the stories your family shares, and the culture of your community. Discover it and improve your health!”

– Sharon F. Terry President & CEO Genetic Alliance

Carlo's Story

How can family health history affect my health?

Every man in my family has dropped dead before the age of 50—my father, my grandfather, and my uncles. I always assumed that I wouldn't make it to 50.

A few years ago my cousin told me that he told his doctor about our family health history.

You inherit many things from your parents and grandparents. They pass on culture and values through photos, recipes, stories, spiritual practices, and music. You also inherit how you look—for example, how tall you are and the color of your eyes. Small structures in cells called genes carry information for these characteristics and how your body works. Your genes were passed on to you from your parents.

Some genes can make it more likely that you will get certain diseases. When members of your family share health problems, you might be at risk for getting the same health problems in the future. This is because family members can have genes, lifestyle, and environment in common. However, you may be able to prevent illness by knowing your family health history and by making healthy choices.



His doctor did some tests and found out that my cousin was at risk for heart disease—maybe even a heart attack.

He put my cousin on medication to lower his cholesterol and told him to stop eating so many fried foods.

I talked to my doctor and got the same tests and advice. Last year, I threw the biggest 50th birthday party ever!

Kathleen's Story

I've always known that some of the relatives on my father's side of the family died in early adulthood. At 23, I started to experience heart problems. I worried that I would die young like my aunt Kathleen, after whom I was named. Like many of my relatives who died, I was unusually tall, and everyone said I was Aunt Kathleen's "spitting image."

I was in medical school at the time and drew a family tree. When I shared my family health history with a few specialists, they did some tests and diagnosed me with Marfan syndrome. I contacted all of the members of the family who I feared were affected, and they were able to get lifesaving treatment.

Now I don't live in fear that I might die suddenly like my aunt. I don't worry for my daughter Marissa, who also has Marfan syndrome. We take the proper medication, get checkups regularly, and can get on with our lives. Our ancestors did not die in vain. Their story was heard and saved many.

To find out more about Marfan syndrome, visit www.marfan.org.



How can my choices affect my health?

Many things shape your health. Some things—such as your genes—are outside of your control. Other things—such as what you eat, if you smoke or exercise, and what you do for a living—can be influenced by the choices you make. To make healthy choices, you need to understand your current health, your risk for getting certain diseases, and your environment.

Family health history is the first step on the road to better health.

What information should I collect?

Who to collect information on:

- Yourself
- Your parents
- Your brothers and sisters
- Your children

Then move on to aunts, uncles, cousins and grandparents.

Basic information to collect:

- Name and how you are related (myself, parent, child, etc.)
- Ethnicity, race, and/or origins of family
- Date of birth (or your best guess—for example “1940s”)
- Place of birth
- If deceased, age and cause of death

Collect stories about your ancestry and culture. This is a great chance to preserve your family’s memories.



Be sure to record age at onset of symptoms

(when the disease started)

Health history:

- Alzheimer's disease
- Asthma and allergies
- Birth defects (such as cleft lip, heart defects, spina bifida)
- Blindness/vision loss
- Cancer (such as breast, ovarian, colon, prostate)
- Current and past medications
- Deafness/hearing loss at a young age
- Developmental delay/ learning disorders
- Diabetes/sugar disease
- Heart disease
- High blood pressure
- High cholesterol

- History of surgeries
- Immunizations
- Mental health disorders (such as depression, schizophrenia)
- Obesity
- Pregnancy (such as number of children, miscarriages, complications)
- Stroke
- Substance abuse (such as alcohol, drugs)

Lifestyle:

- Exercise
- Habits (such as smoking, drinking, regular doctor/ dentist checkup)
- Hobbies and activities
- Nutrition and diet
- Occupation



How do I collect family health history?

Talk to your family

Your relatives are the best source of information about your family. Family history is often shared while talking at family events like birthday parties, weddings, reunions, religious gatherings, holiday dinners, and funerals. These events provide a chance to ask family members about their lives.

Use what you have

Check first to see if your family has family trees, charts, or listings of family members. This information may be written in baby books, photo albums, birthday date books, a family bible or other religious records. Review your own medical history with your doctor to make sure you are not forgetting anything.

Plan an individual conversation

After you have brought up the idea of collecting your family health history, you may want to talk with certain family members to get a more complete record of what they know. If possible, record these talks so you can go over them later. This guide includes questions to ask.

Consider explaining to family members that you are collecting the information to learn more about possible medical conditions you or your children might have in the future.

Send a survey

You may wish to send out a survey asking for health information from relatives. One quick and easy way to collect this information is to put it in a holiday newsletter. Not everyone will feel comfortable filling out a survey. Be sure to explain exactly why you are asking questions.





Lisa's Story

When I became pregnant, I talked with my parents about diseases that run in our family. I wanted to be prepared for anything that could affect my child. My parents told me that, before I was born, my aunt had a baby boy who seemed healthy. A few weeks later, however, he became very ill with vomiting and wasn't eating. The doctor diagnosed him with a genetic disorder called MCADD (Medium Chain Acyl-CoA Dehydrogenase Deficiency) and put him on a special diet and medication. Unfortunately, some damage had already been done.

Tips for collecting family health history

- Start with your parents if they are still living. Often, older relatives are good sources of information and can be the “family historian.”
- If you are adopted, you may be able to learn some of your family history through your adoptive parents. You may also ask to see the adoption agency records.
- It is important to respect others. Some relatives may not want to share their medical histories. Some may not know their family history.
- It can be scary to find out about a health concern in your family. Sharing family history with your healthcare provider can help you understand if you are at risk.
- Family members may not clearly identify all diseases. For example, someone who suffered from “the blues” may have had depression. Ask family members to talk about how relatives acted.

How do I ask my family members about family history?

On the next page is a list of questions that will help you talk with your family members. These questions will help you learn about your family stories, as well as health patterns and any impact environment, lifestyle, and family history may have on family health. Add your own questions that relate to your family.

Prepare ahead of time

- Write down what you know—such as family members' names, where they were born, or how many children they have.
- Pick the questions you will ask ahead of time.
- Record the interviews on a tape recorder or video camera.

When I told my doctor this, she checked to see if MCADD was included in our state's newborn screening program. Luckily, it was. When my daughter was born, we paid close attention to her newborn screening results. She too has MCADD, but we were able to start treatment right away. Now she is a healthy, active three-year-old. By learning about my family health history, I was able to ensure that my baby stayed as healthy as possible from the moment she was born.



To find out more about MCADD, visit www.fodsupport.org.



During the conversation

- Write down health-related information given by your relative.
- Try to keep the questions short. Avoid questions that can be answered with a simple “yes” or “no.”
- Use follow-up questions such as “why,” “how,” and “can you give me an example.”
- Do not expect people to know the answers to all of your questions.
- Respect a person’s wishes not to talk about certain topics.
- Ask your family member to show you pictures, recipes, letters, and other family keepsakes. These can help people remember details and lead to more family stories.



Michael's's Story

Since I am adopted, I thought there was no way for me to put together a family health history. Even though my adopted family and the adoption agency tried to help, they didn't have the answers I needed about my birth family's health. How can I have a family health history without having my questions answered?

There is something I can do. I am starting my family health history with me and my health issues—like the fact that I started wearing glasses when I was 12 years old. I will pass the information on to my children, and they will be able to add to our family history.



ORGANIZE

These questions are examples. You should change them to fit your conversation.

Questions about childhood

- Where were you born?
- Where did you grow up?
- Did you experience any health problems (for example, allergies) as a child?
- Do you have any brothers or sisters? Are they living? How old are they?
- Questions about adulthood
- What jobs have you had? Can you tell me about a normal day?
- What was your work environment like?
- Do you have children? What are their names? When were they born? Did they have any health problems?

- What habits (sun exposure, physical activity, smoking, etc.) have you had that could have affected your health?
- Did you have any health problems as an adult? At what age? How was this treated (e.g. medicine, surgery)?

Questions about parents and grandparents

- When and where were your parents born? What do you know about them (for example, their jobs and hobbies)?
- When and where were your grandparents born? What do you remember about them?
- Did your parents or grandparents have any health problems?
- Do you know if your parents or grandparents took medicine on a regular basis? If so, for what? Did they use home remedies? What kinds and for what?

Questions about family life

- Has your family lived anywhere that caused them health problems (e.g. disaster areas, waste sites)?
- What foods does your family usually eat? Describe a typical family breakfast or dinner. Do you eat special foods for special occasions?
- Has anyone had problems in pregnancy or childbirth? What kinds of problems?
- Are there any diseases that you think might run in our family?
- Is there anything else you would like to tell me about your life or health concerns in our family?

How can I organize my family health history?

The family health history information you collect can be written down or typed into the computer. It is important to write down all of the information so it makes sense to you, your family, and your healthcare provider. We have included some ideas below to help you organize your information.

Family health portrait

A family health portrait is like a family tree showing family members and their health. Because it is a simple picture of your family health history, it can be easily shared with your healthcare provider. Turn to page 18 for more information.

Healthcare Provider Card

Online (www.geneticalliance.org/ccfhh), you will find a card to fill out and bring to your provider. The card focuses on concerns you have about your family health history. It also gives your provider more information on how to best use your family health history to figure out your risk for getting a disease.





Isabel's Story

My grandmother died of a massive heart attack when she was 39 years old. Even though my mother was so careful about healthcare for our family, she did not know that she had a very serious heart condition. She felt just fine, but during a routine checkup two years ago, a doctor discovered my mother's uncontrollable heart arrhythmia. Mom was taken to the hospital immediately, and a cardiac pacemaker was implanted the following day. Since the doctors knew my grandmother had died of a heart attack at a young age, they took my mother's symptoms very seriously.

This knowledge about my mother's and grandmother's health scares me, but I feel it is good to know. I know you can feel healthy but have a serious health condition. Whenever I go for my checkups, I always talk to my doctor about my grandmother's and mother's heart

conditions. She checks my heart thoroughly and always runs a special test on it because of my family health history.

Though I can't change what happened to my mom and grandmother, I can use the information to manage my health better and lower my risk of heart disease.





UNDERSTAND

How can I draw a family health portrait?

If you have access to the Internet, you can use the U.S. Surgeon General's My Family Health Portrait to create a family health history tree on your computer.

Visit www.familyhistory.hhs.gov.

You can also draw your own family health portrait. Use the example on the next page to help guide you.

Instructions for drawing a family health portrait

- Write your name and the date at the top of a large piece of paper.
- Draw yourself at the center using a square if you are a man or a circle if you are a woman.
- Draw your parents above you and label each symbol with his or her name and birth date (or approximate age).

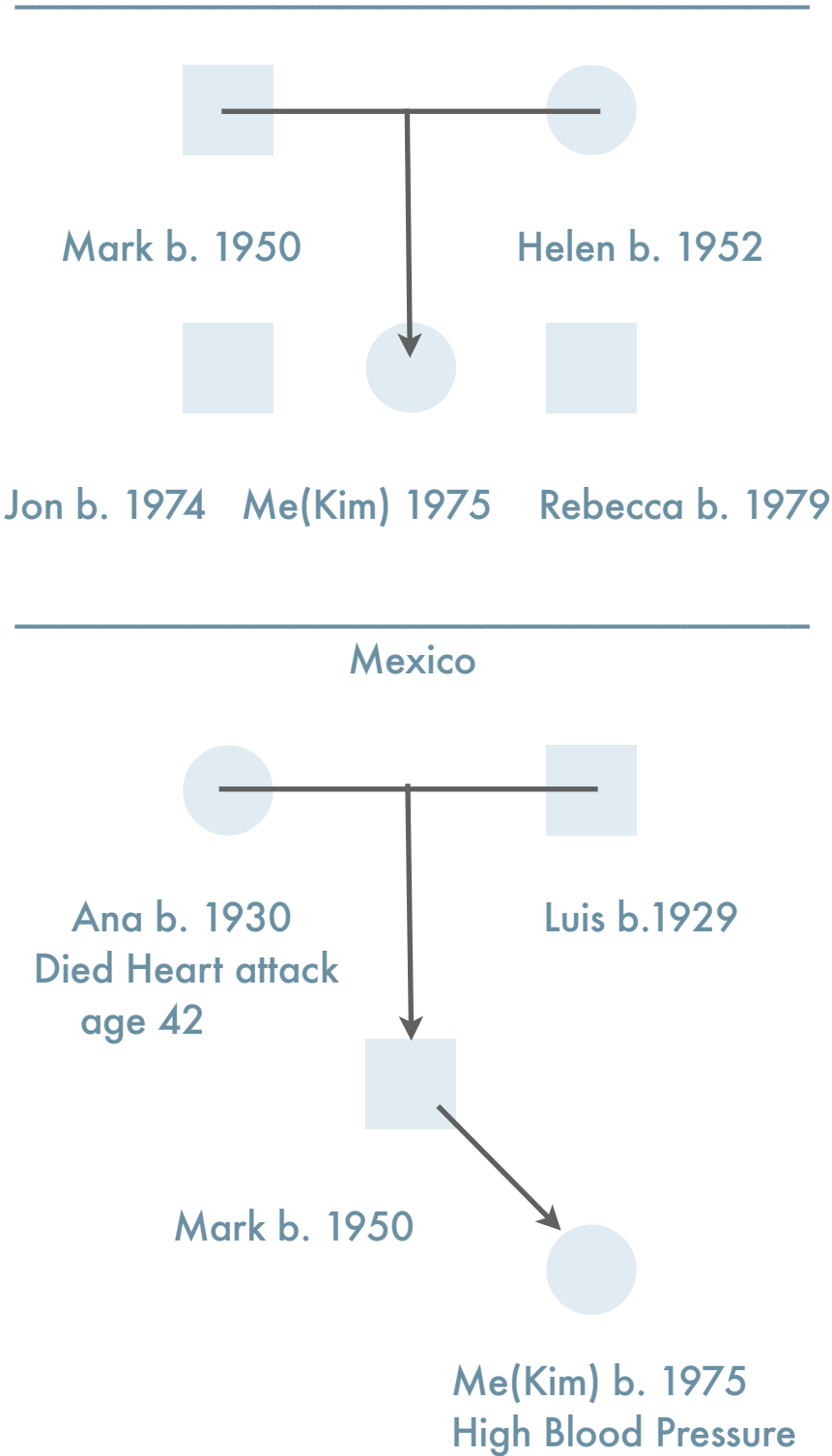
- Draw a line between them and then draw a line down to you.
- When possible, draw your brothers and sisters and your parents' brothers and sisters from oldest to youngest, going from left to right across the paper.
- Add the health information you collected for each individual.
- Add the ancestry and any other information you have collected.

Kim’s Story

1. To start my family health portrait, I added myself. Any women on the portrait are drawn with a circle. Under my name, I wrote my birth date.
2. Next, I drew in my mother and father. Any men in the portrait are drawn as squares. I connected my parents to each other using one line and drew another line that connected to me.
3. Next, I added my brother and sister and their birthdates. I connected each of them to the line that connects my parents.
4. For my father’s parents, I put their names above his and drew one line connecting them to each other and another line connected to my father. I also added my family’s ancestry. Finally, I drew a line diagonally through my grandmother’s circle to show that she died at age 42.



Me (Kim) b 1975





What now?

- Save your family health history and update it with new information you learn about your family.
- Find out more about the diseases that run in your family and how to stay healthy.
- Share information with your family.
- Take your family health history to your healthcare provider.

Resources

Genetic Alliance Family Health History Resources

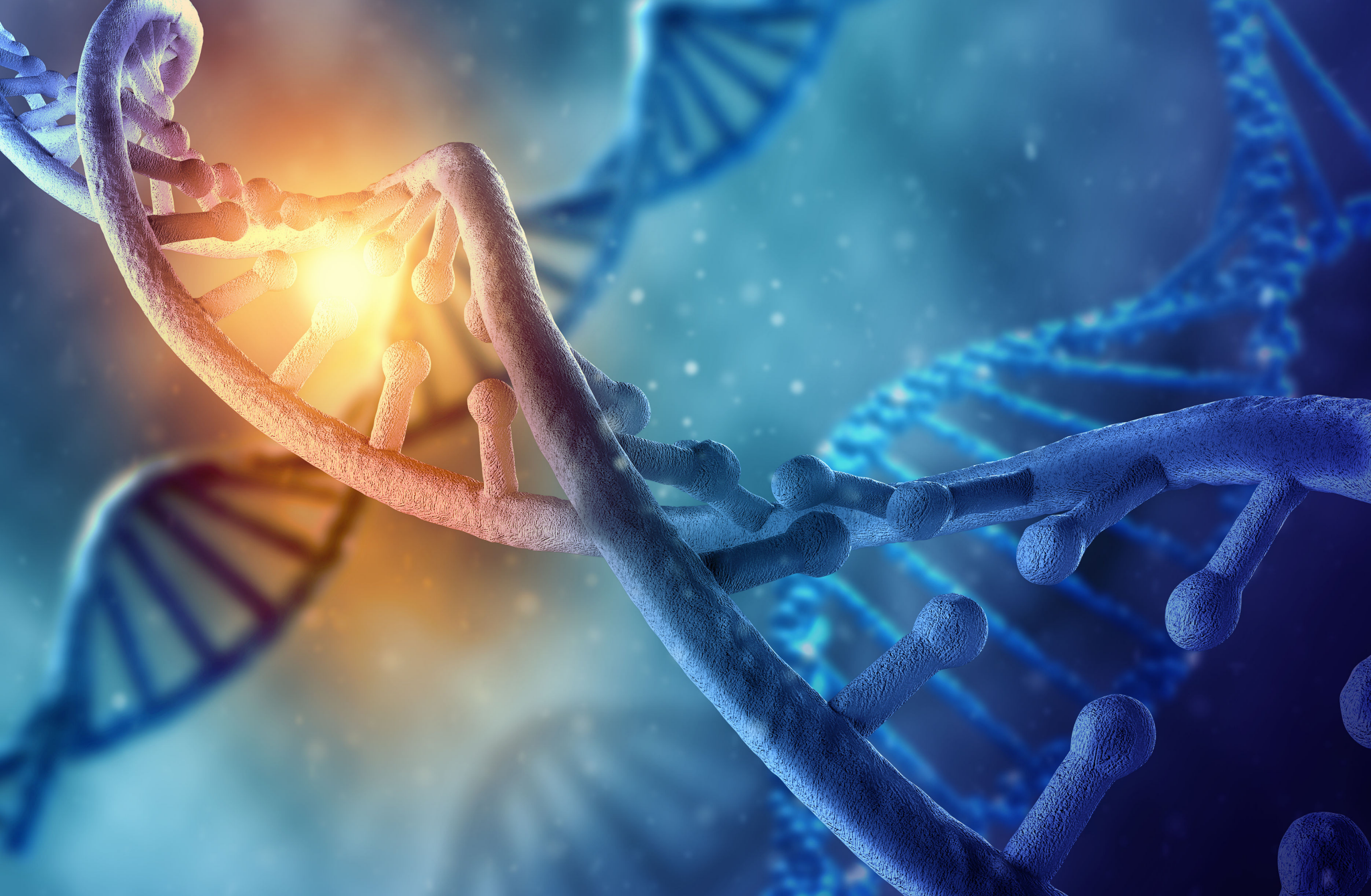
www.geneticalliance.org/familyhealthhistory

The Smithsonian Folklife and Oral History Interviewing Guide
www.familyoralhistory.us/news/view/the_smithsonian_folklife_and_oral_history_interviewing_guide/

and visit the American Folklife Center website:

www.loc.gov/folklife/fieldwork

Free Patient Education from Lucid Genetics



2

GENETICS AND HEALTH

Why is genetics important to my family and me?

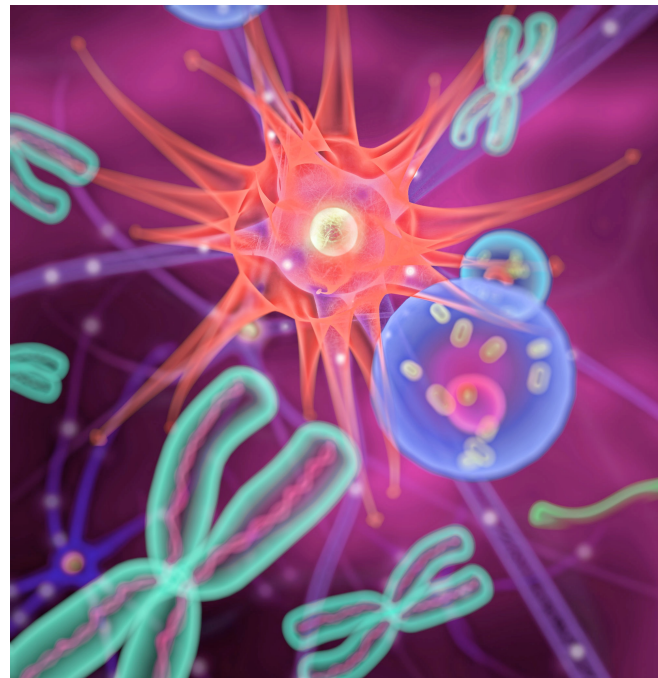
Genetics helps to explain:

- What makes you unique, or one of a kind
- Why family members look alike
- Why some diseases like diabetes or cancer run in families
- How learning your family health history can help you stay healthy
- Why you should bring your family health history to your healthcare provider

Taking time to learn about health and diseases that run in your family is worth it! It will help you understand your own health and make healthy choices.

What makes me unique?

Every person is unique. Part of what makes you unique is your genes. Genes are the instructions inside each of your cells. They control how you look and how your body works. Since everyone has slightly different genes, everyone has a different set of instructions. Genes are one reason why you are unique!



Tell me more about my genes

- A person has two copies of each gene, one from the mother and one from the father.
- Genes carry instructions that tell your cells how to work and grow.
- Cells are the building blocks of the body. Every part of your body is made up of billions of cells working together.
- Genes are arranged in structures called chromosomes. Humans have 23 pairs of chromosomes. Copies of the chromosomes are found in each cell.
- Chromosomes are made up of DNA. DNA is the special code in which the instructions in your genes are written



Why do family members have things in common?

You share a lot with your family—including what can make you sick.

Common Disease: Diabetes

Changes in your genes passed on by your parents may make you more likely to develop type 2 diabetes. If you are active and eat a healthy diet, you may be able to lower your risk.

Children inherit pairs of genes from their parents. A child gets one set of genes from the father and one set from the mother. These genes can match up in many ways to make different combinations. This is why many family members look a lot alike and others don't look like each other at all. Genes can also increase the risk in a family for getting certain health conditions.

Families also share habits, diet, and environment. These influence how healthy we are later in life.

Why do some diseases run in families?

Some diseases are caused when there is a change in the instructions in a gene. This is called a mutation. Every person has many mutations. Sometimes these changes have no effect or are even slightly helpful. But sometimes they can cause disease.

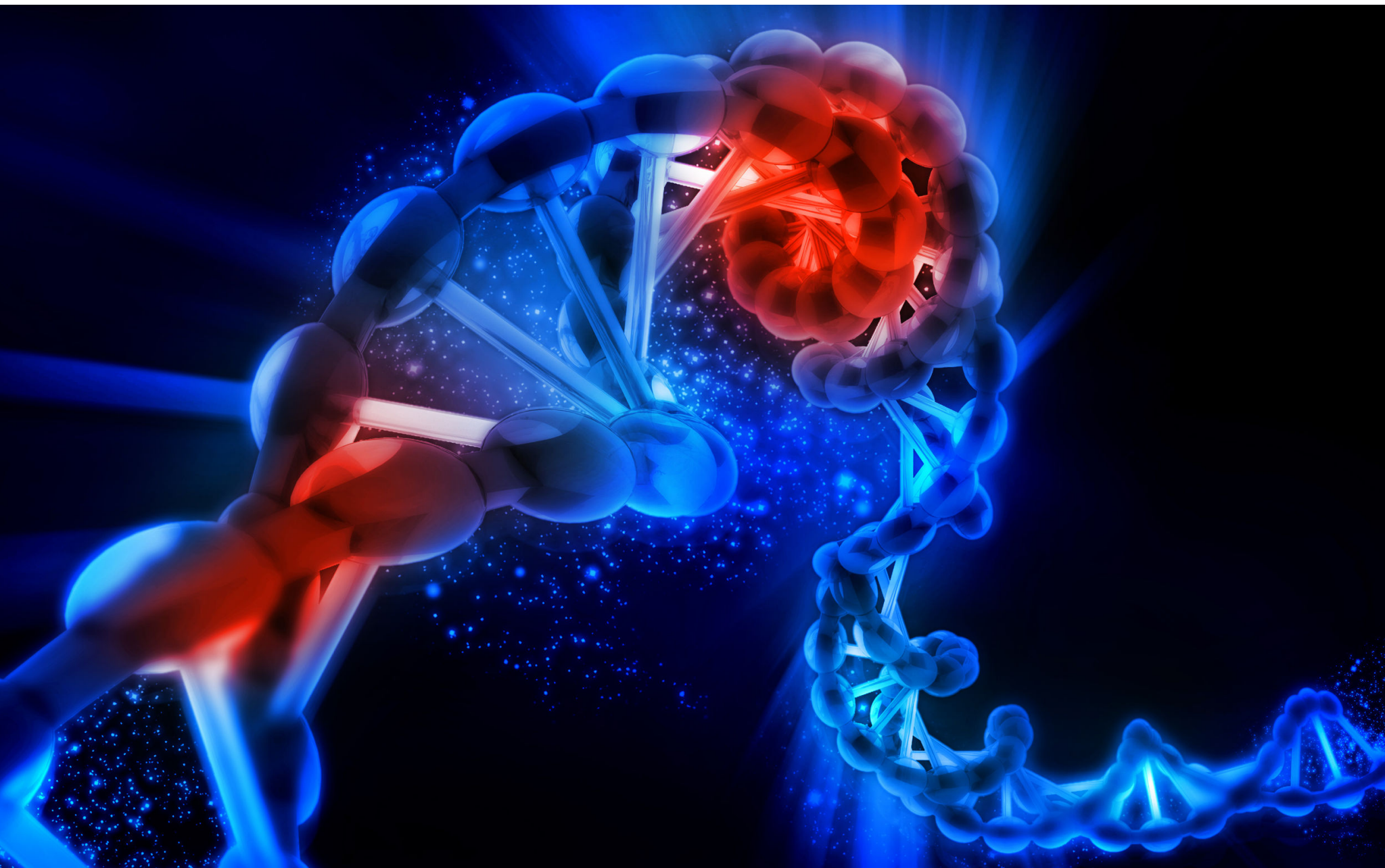
Most common diseases are caused by a combination of mutations, lifestyle choices, and your environment. Even people with similar genes

may or may not get an illness if they make different choices or live in a different environment.

Thousands of diseases are caused by a specific change in the DNA of a single gene. Many of these diseases are rare. These conditions usually develop when an individual is born with a mutated gene.

If a rare disease runs in your family, be sure to write it down. Do not forget to learn about common conditions that affect your family's health.

Single Gene Disorder:
Sickle Cell Anemia
Sickle cell anemia is caused by a mutation in a single gene passed from each parent.



Remember

1. Share your family health history with your healthcare provider.
2. Ask if you can be screened for a disease that runs in your family.

How can knowing my family health history help me stay healthy?

Your family health history tells you which diseases run in your family. Health problems that develop at a younger age than usual can be a clue that your family has a higher risk. Though you cannot change your genes, you can change your behavior.

Knowing your family health history will help you:

- Identify risks due to shared genes.
- Understand better what lifestyle and environmental factors you share with your family.
- Understand how healthy lifestyle choices can reduce your risk of developing a disease.
- Talk to your family about your health.
- Tell your healthcare provider about diseases that run in your family.

Why should I take my family health history to my healthcare provider?

Your healthcare provider (doctor, nurse, or physician's assistant) may use your family health history and current health to figure out your risk for developing a disease. Your provider can then help decide which screenings you get and which medicines you might take.

Based on your family health history, a healthcare provider may order a genetic test or refer you to a genetic counselor or geneticist. Genetic tests can show if you have a gene change that increases your risk for disease. They can also tell if you have a gene change that you might pass on to your children. Your healthcare provider can help you:

- Understand the results of your tests.
- Learn of any treatments for a disease found by the test.

All newborn babies born in the U.S. and many other countries are tested for certain genetic diseases that may make them sick. This is called newborn screening. If the screening test finds a problem, a healthcare provider will help you understand what can be done to help the baby.



DISEASES THAT RUN IN THE FAMILY

In the rest of this booklet, we give you examples of some common diseases that affect our communities and families. For each disease, we include information under the following headings:

- What is the disease?
- Who is at risk?
- Hints for health

For more information, visit www.nhlbi.nih.gov/health/dci and click on “Coronary Artery Disease” or call the American Heart Association at 800-AHA-USA-1 (800-242-8721).

Heart disease

Heart disease is the main cause of death in America in both men and women. There are many types of heart disease. Two of the most common types are coronary artery disease (CAD) and high blood pressure (hypertension).

What is Coronary Artery Disease (CAD)?

- In CAD the arteries that supply blood to the heart muscle can get hard and narrow. The arteries narrow, or get smaller, because plaque and cholesterol build up on the inner walls.
- CAD gets worse over time. As the arteries get smaller, less blood gets to the heart, and less oxygen gets to the heart muscle. Very low levels of oxygen can cause chest pain or a heart attack.
- CAD is the most common cause of heart attacks among Americans.

Who is at risk?

- About 13 million Americans have CAD.
- Everyone has some risk for developing heart disease.
- CAD is caused by a combination of genes, lifestyle choices, and environment.
- For some people, a healthier diet and more activity can change cholesterol level and lower risk.
- Since your genes cannot be changed, some people need medicine to lower their risk of having a heart attack.

Hints for health

- Eat healthy meals.
- Get active and exercise regularly. Obesity increases your risk.
- Take your prescribed medications to control high cholesterol, high blood pressure, and diabetes.
- If you smoke, talk with your healthcare provider about quitting.

What is High Blood Pressure?

- Blood pressure is a measure of how hard your heart is working to push the blood through your arteries, the blood vessels leaving your heart.
- There are two numbers in a blood pressure reading. A normal reading is about 120/80 (read as “120 over 80”). The first number is the force your heart uses to pump the blood. The second number is the pressure between heartbeats.
- High blood pressure means that your heart is working too hard. Over time, high blood pressure can cause kidney failure, heart attacks, strokes, and other health problems.

Who is at risk?

- About one in three adults has high blood pressure. Many do not even know it because there are no clear symptoms.
- A family history of high blood pressure increases your risk for getting it at a younger age.
- Risk increases with age, being overweight, or having a family history of hypertension.

Hints for health

- Eat less salt.
- Maintain a healthy weight.
- Manage your stress.
- Get active and exercise regularly.
- Limit the alcohol you drink.
- Get screening regularly.

For more information, visit www.nhlbi.nih.gov/health/dci and click on “High Blood Pressure” or call the American Heart Association at 800-AHA-USA-1 (800-242-8721)

Heart disease symptoms may not appear until the damage is already done. Talk to your family about heart disease today.

For more information, visit www.nhlbi.nih.gov/health/dci and click on "Asthma" or call the American Lung Association at 800-548-8252.

What is Asthma?

- Asthma is a lung disease that causes repeated episodes of breathlessness, wheezing, coughing, and chest tightness. The episodes can range from mild to life threatening.
- Asthma episodes are caused by triggers. These are things like dust mites, animal dander, mold, pollen, cold air, exercise, stress, viral colds, allergies, tobacco smoke, and air pollutants.
- Some people have genes that control their response to these asthma triggers.

Who is at risk?

- Asthma affects about one in 10 children and one in 12 adults.
- Asthma is the main reason children end up in the emergency room and miss days of school.
- If you have parents, siblings, or children with asthma or allergies, you are more likely to get it.

Hints for health

- Avoid exposure to triggers.
- Use medication correctly.

Diabetes (sugar disease)

Diabetes is a serious, chronic disease in which blood sugar levels are above normal. Many people learn about their diabetes after problems develop. According to the American Diabetes Association, one out of three people who have type 2 diabetes do not know that they have the disease.

Symptoms occur when the body fails to change sugar and other food into energy. This happens when the body cannot make or use a hormone called insulin. Serious problems from diabetes can include blindness, kidney failure, and death. Diabetes can be detected early and treatment can prevent or delay these serious health problems.

For more information, visit www.ndep.nih.gov or call 800-860-8747.



Both genetic and environmental factors such as diet and exercise plays an important role in getting the disease.

What is Type 1 Diabetes?

- Type 1 diabetes usually develops in young children or young adults.
- People with type 1 diabetes stop making their own insulin.

What is Type 2 Diabetes?

- Type 2 diabetes usually develops in people over 30 years of age. In recent years, more young people are getting it due to poor diet.
- Scientists are learning more about the specific genes involved in this type of diabetes.
- Who is at risk?

- Diabetes affects about one in 13 people in the United States.
- Five to 10 percent of Americans with diabetes have type 1 diabetes.
- Children or siblings of people with diabetes are more likely to get diabetes.
- Obese people are more likely to get type 2 diabetes.
- Women who had a baby that weighed more than 9 pounds or who had gestational diabetes while pregnant are at risk.
- Hints for health
 - Eat more fruits and vegetables, less sugar and fat.
 - Get active and exercise regularly.
 - Lose weight if necessary.

Cancer

There are many types of cancer. Cancer is caused by the growth and spread of abnormal cells. Though your risk of getting cancer increases as you get older, genetic and environmental factors also cause people to be at a higher risk for certain types of cancer. Some of the most common cancers are breast cancer, lung cancer, and prostate cancer.

What is Breast Cancer?

- Breast cancer is a type of cancer that forms in the tissues of the breast, usually the ducts.
- Breast cancer is one of the most common cancers among women.
- Although it is rare, men can also get breast cancer.

For more information, visit www.cancer.gov/cancertopics and click on “Breast Cancer” or call 800-4-CANCER (800-422-6237).



- Most breast cancer can be treated if found early.

Who is at risk?

- One out of eight American women will develop breast cancer in her lifetime.
- Among Hispanic/Latina women, breast cancer is the most common type of cancer.
- Breast cancer risk is higher if a woman has close blood relatives who have had this disease. Both your mother's and father's family history of breast cancer is important.

Hints for health

- Women should do monthly breast self-exams.
- After age 40, women should get annual mammograms.
- Ask about genetic testing for high-risk families.
- Eat a healthy, balanced diet.
- Get active and exercise regularly.
- Limit the alcohol you drink.

What is Lung Cancer?

- Lung cancer is the uncontrolled growth of abnormal cells in one or both of the lungs.

Who is at risk?

- Lung cancer is the leading cause of cancer death for both men and women.
- About 160,000 people died in the United States from lung cancer in 2007.
- Nearly 87 percent of lung cancer cases in the United States are smoking-related.

Hints for health

- Do not smoke.
- Avoid secondhand smoke.
- Find out about testing for radon and asbestos in your home and at work.

What is Prostate Cancer?

- Prostate cancer develops in the male reproductive system. The prostate is a small gland near the bladder.
- Scientists do not yet know what causes prostate cancer.
- Doctors have a test to find out whether a man might have prostate cancer.

Who is at risk?

- Men of all ages can develop prostate cancer. However, more than eight out of 10 cases occur in men over the age of 65.
- Prostate cancer is the most common type of cancer diagnosed in Hispanic/Latino and African American men.
- Having a father or brother with prostate cancer more than doubles a man's risk for getting prostate cancer. The risk goes up with the number of relatives who have it, especially if the relatives were less than 50 years old when they got it.

Hints for health

- Follow a healthy diet.
- Exercise regularly.
- After age 50, have your prostate checked.



SINGLE GENE DISORDERS

What are Single Gene Disorders?

- Earlier in this booklet, you read about conditions caused by mutations in a single gene. These conditions are called single gene disorders.
- There are more than 6,000 single gene disorders. Combined, they occur in about 1 in 300 births.
- The symptoms of single gene disorders vary widely, but many of them run in families.
- Collecting your family health history for these conditions is important for diagnosis and management of the condition and for making reproductive choices.
- Who is at risk?
- Every person is born with mutations. Most of these mutations will not cause disease on their own, but it is important to identify any that do.

- Depending on which gene is affected, single gene disorders can be passed down even when the mother and father do not show any symptoms.
- Some single gene disorders are identified during a pregnancy or soon after a child is born. Others will not be diagnosed until adulthood.
- The most harmful mutations may lead to a miscarriage or stillbirth. If you have a family history of miscarriages, this may be related to a genetic mutation.



Picture courtesy of CureAHC Inc.



Hints for health:

- If you have a family history of a single gene disorder, discuss it with your healthcare provider. Your provider may refer you to a specialist.
- Know which newborn screening tests are performed in your state.
- For thousands of conditions, advocacy organizations provide support services, information, and ways to get involved in the discovery of treatment options.

Visit Disease InfoSearch at www.geneticalliance.org to find out more.

For more information on single gene disorders, contact the Genetic and Rare Diseases (GARD) Information Center at gardinfo@nih.gov or 888-205-2311.

Resources

The “Does It Run In the Family?” toolkit includes two pieces that can help you summarize your health information for your provider—the family health portrait and healthcare provider card. You may also hear your healthcare provider call a Family Health Portrait a “pedigree.”

Each family and individual is unique and may have genetic diseases other than the major diseases listed here.

For more information visit:

Disease Info Search

www.geneticalliance.org

National Library of Medicine

www.nlm.nih.gov/services/genetics_resources.html



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Genetic Alliance transforms health through genetics. We promote an environment of openness centered on the health of individuals, families, and communities. We bring together diverse stakeholders that create novel partnerships in advocacy; integrate individual, family, and community perspectives to improve health systems; and revolutionize access to information to enable translation of research into services and individualized decision making.



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